Original Article

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Study of Congenital Malformations in Central Nervous System in Newborns

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Abstract

Introduction: Among all the congenital anomalies, disorders of the CNS are the most severe, difficult to understand its etiology, and predict its clinical presentation and course. 75% of fetal deaths and 40% of deaths within the first year of life are secondary to CNS malformations. **Aims & Objectives:** To study the incidence, pattern and outcome of CNS anomalies in newborns. To established the possible etiological factors and relationship of various antenatal factors. **Materials & Methods:** All babies were examined within 24hrs of birth & followed up to 72hrs, any anomaly detected confirmed by required investigations. **Study Period and design:** January 2012 to June 2013. Cross-sectional study (prevalence study). **Statistical analysis:** Analyzed by simple statistical techniques and tests of significance including Chi-square tests were applied. **Results:** There were total of 10294 newborn of which CNS malformations were 151 patients with 188 anomalies(1.46%). Most frequent CNS malformations was anencephaly 55/188 (29.25%) followed by Hydrocephalus 51/188 (27.13%), spina bifida (19.15%), Meningocele/meningomyelocele (12.77%), encephalocele (3.19%), agenesis of corpus callosum (3.72%). Male were most commonly involved than female (84% v/s 67%). 107/151 (70%) mother did not have preconceptional and antenatal folic acid supplementation. 15/151 (30%) mother had antenatal history of anaemia. Incidence was more in maternal age > 30 years and multiparity. Incidence of congenital anomalies was more in preterm (41.8/ 1000) as compared to full term (10/ 1000). 50 newborns (33.11%) expired within few hrs. **Conclusion:** Maternal age and parity are important risk factor andpreterm and low birth weight babies are at high risk of CNS Malformations.

Keywords: Congenital central nervous system; Malformations; Newborn; Prevention; Prenatal diagnosis.

Introduction

Congenital malformations are a major cause of prenatal and neonatal death, both in developed and developing countries. The field of dysmorphology has expanded dramatically as the number of recognizable patterns of malformations has more than tripled during the last 25 years.[1]

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These malformations have multifactorialetiologies and 40% of cases are idiopathic, but there is an impression that they are more prevalent in populations with consanguineous marriages.[2] Genetic and congenital diseases are almost always serious, incurable, a number of these diseases are treatable, and in some cases, their clinical therapeutic intervention and study of family history and genetic counseling remains of paramount importance.[3]

Congenital malformation will begin to emerge as one of the major childhood health problems. Treatment and rehabilitation of children with congenital malformations is costly and complete recovery is usually impossible. Approximately, 66% of major malformations have no recognized etiology

and most of them have multifactorial inheritance. These defects can occur for many reasons including inherited genetic conditions, poor diet and toxin exposure of the fetus for example to alcohol, birth injury and in many other cases for unknown reasons.[4]

There are several reports that suggest that the incidence, and particularly the pattern, of congenital central nervous system (CNS) anomalies may vary in different geographical locations. However, the extent to which such reported variations are attributable to differences in genetic predisposition, environmental factors or diagnostic precision is uncertain. Studies on the incidence and pattern of different types of congenital abnormalities can provide valuable information for planning health care services, including preventive programs.[5]

The present study is aimed at analyzing the incidence and pattern of congenital CNS malformations and relation to various antenatal factors in the newborns in tertiary health care centre so that future preventive strategies are planned.

Material & Methods

Study was carried out in GMC Latur for a period of 18 months from January 2012 to June 2013. All live newborns and stillborns were included in this study where as all babies born outside, referred to NICU and Abortions (Gestational age <28wks) were excluded from the study.

Data collection was performed in two parts. At first part, variables recorded were about maternal details and included the date of admission, age, gravida, parity, history of chronic illness, drug ingestion, exposure to radiation, history of congenital malformations in other offspring, parental consanguinity, nutritional status of mother, history of smoking, tobacco chewing & alcohol consumption, paternal and maternal occupation were obtained. Certain fatal and placental conditions like APH, placenta previa, hydramnios were noted. The second part was about neonatal details including live or stillbirth, gestational age, birth order, sex, existence of Central nervous system congenital anomaly and type of it, any investigation done. No autopsy examinations were performed. All babies were examined within 24 hrs of birth & followed up upto 72 hrs. The gestational age of baby was assumed from examination of baby and the age previously calculated from Last Menstrual Period was confirmed. Birth weight, sex of each baby was noted and detail physical examination was done. The study cases were investigated by means of X-ray, CT and MRI were done in those cases where diagnosis was inconclusive on clinical examinations. All details were recorded in predesigned patient information sheet after taking written informed consent. Observations were tabulated and analyzed. Statistical analysis was done using Chi-square test.

Observations & Results

Total no. of newborns delivered was 10294 (live births 9861+still births 433) of which 151 (1.46%) newborns had single or multiple CNS malformation (27 still births + 124 live births). I ncidence of CNS anomalies was 14.67 per

Chart 1: Relation of Maternal age with congenital malformations

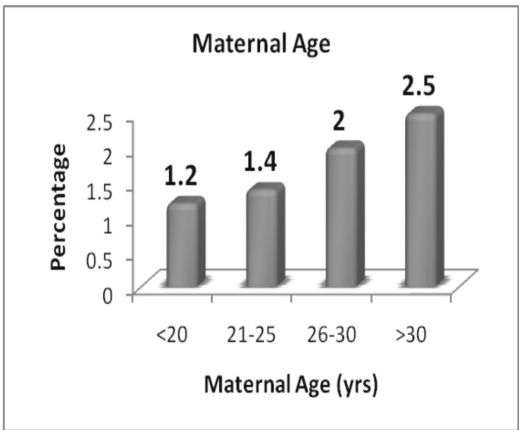


Table 1: Relation of parity with congenital malformations

Parity of mother	No. of cases	Total no. of deliveries	Percentage
1	61	4614	1.3
2	42	3060	1.4
3	33	1699	1.9
4	10	417	2.4
>5	5	200	2.5
Total	443	9990	

Table 2: Correlations of antenatal factors with congenital anomalies

Maternal factors	Percentage
Folic acid supplementation not taken	70
Anaemia	31
Hydromnios	16
Previous abortion	14
Drugs/alcohol/tobacco	12
Fever during first trimester	11.5
Pre-eclamptictoxaemia	10.38

1000 births. Incidence in live birth was 12.57 per 1000; while incidence in still birth was 62.36 per 1000. Only 17 neonates (11.25%) had parental history of consanguinity. Maximum incidence of congenital anomalies were observed in maternal age group >30 years (chart no.)

Highest incidence of anomaly was observed among newborns of > 4 order of birth (Table 1).

High association of congenital anomalies was noticed with lack of folic acid supplementation during antenatal period (Table 2).

51.69% cases had antenatal detection of anomalies on ultrasonography. It is not done in 42.38% cases but 10% cases had normal antenatal ultrasonography. Incidence of congenital anomalies was more in preterm (41.8/1000) as compared to full term (10/1000) as shown in Table 3.

Incidence of anomalies was more in low birth weight new born (<2500gms) as compared to birth weight >2500 gms (Table 4)

Anomalies were more in males 86 (56.8%) as compared to females 65 (43.11%).Pattern of CNS anomalies found in this study were shown in Table 5.

Most frequent CNS malformations was anencephaly (fig 1) 55/188 (29.25%) followed by Hydrocephalus (fig 2) 51/188 (27.13%), spina bifida 36/188 (fig 3) (19.15%), Meningocele/meningomyelocele (fig 4) 24/188 (12.77%), encephalocele (fig 5) 6/188 (3.19%), agenesis of corpus callosum (3.72%), Dandy walker malformations (1%) and others CNS malformations (3.7%).

Out of 151 CNS anomalies, 27 (18%) newborns were still births. Sixty (40%)

Table 3: Relation of CNS malformation with maturity

Gestation	No. of Cases	Total No. of Births	Incidence/1000
Preterm	62 (41.08%)	1482	41.8
Full Term	89 (58.91%)	8812	10
Total	151	10294	

Chi Square Test=267.514 p<0.001, Highly Significant df=1

Table 4: Association of birth weight with congenital anamolies

Birth weight	No. of cases	%
<2500	90	60
>2500	61	40
Total	151	100

Table 5: Showing pattern of CNS Malformations

Pattern	Total pt. 151) *Total anomalies (188)	%	Incidence/ 1000 Live birth
anencephaly	55	36	5.44
Hydrocephalus	51	34	5
spina bifida	36	24	3.3
Meningocele/ meningomyelocele	24	16	2.13
Encephalocele	6	4.0	0.87
Agenesis of corpus callosum	7	4.6	0.3
Dandy walker malformations	2	1.3	0.2
Other**	7	4.6	0.3

*37 patients have multiple CNS anomalies.** other: aqueduct stenosis, semilobarholoprosencephaly, hypoplasticcerebrum, patau syndrome.

Fig 1: Anencephaly

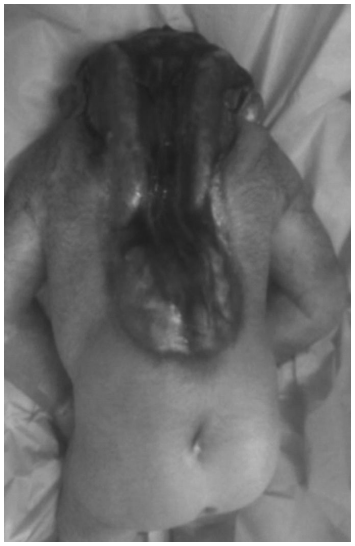


Fig 2: Hydrocephalus with Spinabifida

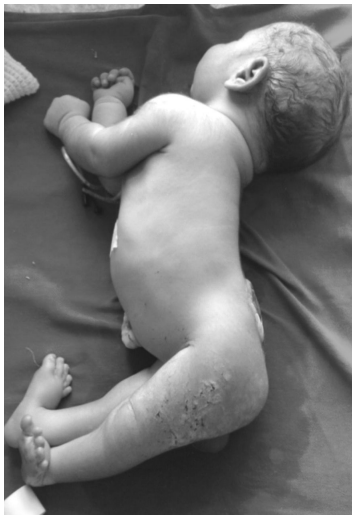


Fig 3: Spinabifida

Fig 4: Occipital Meningocele

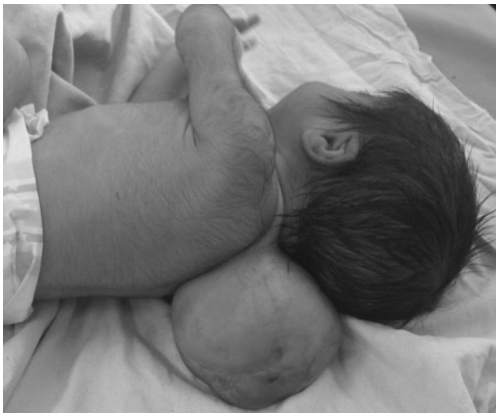
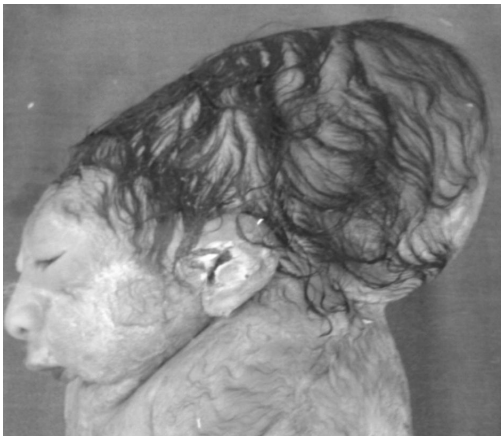


Fig 5: Encephalocele



newborns were expired within 72hrs after birth.(Anencephaly-55, hydrocephalus-2, encephalocele-2, meningocele with multiple anomalies-1). 58 (38%) newborns were referred to different surgical units for correction of anomalies, (Hydrocephalus-27, spinabifida +meningomyelocele-31). Only 6 (4%) newborns were discharged (Agenesis of corpus callosum-4, dandy walker syndrome-1, patau syndrome-1).

Discussion

In present study, the incidence of CNS anomalies was 14.67 per 1000 births Total no. of newborns with CNS anomalies were 151 (1.46%), 27 still births + 124 live births. It is observed that malformations are much more

common in still births 62.36 per 1000 as compared to live births 12.57 per 1000.

Thus, our study found that central nervous system anomalies were the commonest congenital anomalies, which contribute to incidence of 14.96/1000. Our findings are consistent with study of Mital VK *etal[6]* which also showed CNS as the commonest anomaly with incidence of 6.74/1000, Verma M *et al[7]* with incidence of 20.6/1000, Gupta S *et al[8]* with incidence of 6.4/1000.

According to Sayyed SS *et al[9]* CNS malformations were found to be 4.96% and GIT 4.01%. Further it is observed that malformations are much more common in still births (6.53%) as compared to live births (0.63%) similar to this study.

Al-Gazalia, L Sztrihaa, *et al[5]* found that 31 babies had CNS abnormalities giving an incidence of 3.2:1000, The consanguinity level in babies with CNS abnormalities was 62% compared to a consanguinity level of 54% in the general population, but in this study only 17 neonates (11.25%) had parental history of consanguinity. The spectrum of CNS malformations was neural tube defects 49/127 (36.8 % of all CNS malformations), followed by hydrocephalus 34/127 (26.8%). Neural tube defect (NTD) was present in 11 cases (1.14:1000), hydrocephalus in four cases (0.4: 1000) as in our study.

Out of 151 CNS anomalies, 27 (18%) newborns were still births. 60 (40%) newborns were expired within 72 hrs after birth.

Prevention is certainly the best form of therapy. Primary prevention of CNS malformations is limited, with an exception of neural tube defects. Periconceptional folic acid supplementation and/or food fortification with folic acid have reduced significantly both the first occurrence and recurrence of NTDs in the offspring. The consumption of 0.4 mg of folic acid daily is advisable for all women of childbearing age.

There is evidence suggesting that folic acid and other vitamin supplementations prevent neural tube defects and some other form of congenital anomalies. In our studies this may be the risk factor for higher incidence, which requires more extensive study to prove.

Conclusion

- CNS anomalies are one of the most common congenital anomalies with higher incidence of morbidity and mortality.
- Its prevention not only reduces mortality but also morbidity and later handicaps.
- Maternal age and parity are important risk factors.
- Preterm pregnancy and low birth weight babies are at high risk.
- One of the major steps in reducing incidence would be early detection and Medical Termination of Pregnancy.
- Improved antenatal detection & MTP, routine vitamin supplementation specifically folic acid, preconception & for the first 12 weeks of pregnancy should be given emphasis.
- For this, proper antenatal care and high degree of awareness are essential.

References

1. Kliegman RM, Jenson HB, Behrman RE, Stanton BF. Nelson Textbook of Paediatrics, 18th ed. Philadelphia: WB Saunders; 2008, 711-713.
2. Z Mosayabi, AH Movahiden. Pattern of congenital malformation in consangious & non consangious marriages. *Eastern Mediterrian Journal.* 2007; 13(4).
3. MunjalPandya1, Janki Thakkar . Study of Gross Congenital Malformations In Newborn. *Journal of Evolution of Medical and Dental Sciences,* 2013; 2(27): 4989.
4. Dr Akruti Parmar *et al*. A Study of Congenital Anomalies In Newborn. *NJIRM.* 2010; 1(1): 13-18.

5. Al-Gazali LI, Sztriha L, Dawodu A, Bakir M et al. Pattern of central nervous system anomalies in a population with a high rate of consanguineous marriages. *Clin Genet.* 1999; 55: 95–102.
6. Mital VK, Grewal RS. Congenital Anomalies in Neonates. *Indian J Pediat.* 1969; 36: 356.
7. Verma M, Chhatwai J, Singh D. Congenital Malformations-A Retrospective Study of 10,000 Cases. *Indian J Pediatr.* 1991; 58: 245-252.
8. Gupta S, Gupta P, Soni JS. A study on incidence of various systemic congenital malformations and their association with maternal factors. *National journal of medical research.* 2012; 2(1).
9. Saiyad SS, Jadav Hrishikesh R. Study of Congenital Malformations in Central Nervous System & Gastrointestinal Tract. *National Journal of Medical Research.* 2012; 2(2): 1-3.

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